

FIG. 1 Alignment of mouse sequences with the human chromosome 12 genomic clone NT_009720

Expect = 4e-28, Identities = 179/224 (79%), Gaps = 4/224 (1%)

Query: 696 ctttgggacagtgagagctgcctttcatagaaaaatggccttgtgctcctgcttcagcca 755
||||| ||||||||| ||||||||| |||| | ||||||||| | ||||
Sbjct: 315187 ctttgggtgcagtgagagccgcctttcataggaaaacagt-ttgtgctcctgactgggcca 315129

1/15

Query: 756 cctttcaccacctgctcgatt-gcggagcatgtgtgagagg-cagggataaaagggctca 813
||||| |||| | | | | | | | | | | | | | | | |
Sbjct: 315128 ccttcaccccttgttcaagttagcagctcatttgtaaggggtcaggaataaaagggctct 315069

Query: 814 ctctgccctttcccatgtgcaggaagtggccccaggagtgaggagttgtgtccaaaat 873
||| |||| | |||| |||| |||| |||| |||| |||| ||||
Sbjct: 315068 ttcttcctctccatgtgtaggaagtcagcccttgggtgtggagagtcatttctcaaat 315009

Query: 874 aga-cttcctaatacacagttccaaagaggccaagagtcagtca 916
||| ||||||||| ||||||||| ||||||||| |||||||||
Sbjct: 315008 agatcttcctaatatggttccaaagagagcgaagagtcagtca 314965

FIG. 2 The Human RFX4 Locus

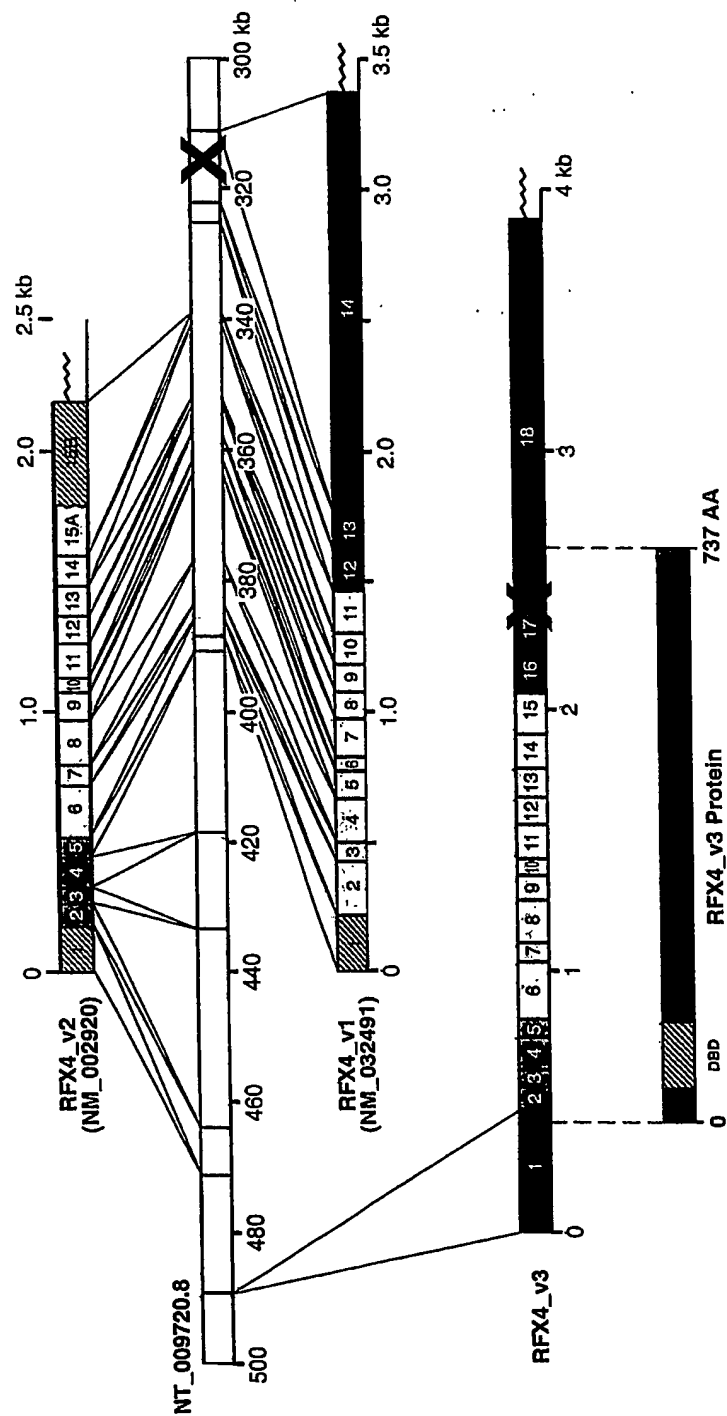


FIG. 3 Alignment of human and mouse proximal promoters for RFX4_v3

Expect = e-107

Identities = 212/216 (98%)

Human 786 gaggggccaacatctaagccaatttttgatttcgcctataatgagtgccggcggaaggctg 845
||||| || ||||||||||||||||||| ||||||||||||||| |||||||
Mouse 86 gagggggaagatctaagccaatttttgatttcgctataatgagtgccggcggaaggctg 145

Human 846 gagaaggcctctggaaactttaataaagaaaacgttgctaataataagaagggga 905
||||| ||||||||||||||||||| ||||||||||||||| |||||||
Mouse 146 gagaaggcctctggaaactttaataaagaaaacgttgctaataataagaagggga 205

Human 906 agtcggagggtgggattgcgtcgctctgagcccccccttttcggaggcggttttcttat 965
||||| ||||||||||||||||||| ||||||||||||||| |||||||
Mouse 206 agtcggagggtgggattgcgtcgctctgagcccccccttttcggaggcggttttcttat 265

Human 966 tcaaaaacaggccccacaatgggcttcac 992
||||| |||||||||||||||||||
Mouse 266 tcaaaaacaggccccacaatgggcttcac 292

FIG. 4 Human, mouse, and zebrafish alignment of RFX4_v3 (amino terminus)

human	MHCGLLEEPDMDSTESWIERCLNESENKRYSSHTSLGNVSNDEEKENNRASKPHSTPA	60
mouse	MHCGLLEEPDMDSTESWIERCLNESENKRYSSHTSLGNVSNDEEKENNRASKPHSTPA	60
zebrafish	MLCGLLEEPDMDSTESWIERCLNESENKRFSSHSSIGNISNDEEKENNRASKPHSTPA	60
	* * * * *	
human	TLQWLEENYEIAEGVCIPRSALYMHYLD FCEKNDTQPVNAASF GK IIRQFFPQLTTRRLG	120
mouse	TLQWLEENYEIAEGVCIPRSALYMHYLD FCEKNDTQPVNAASF GK IIRQFFPQLTTRRLG	120
zebrafish	TLQWLEENYEIAEGVCIPRIALYMHYLD FCEKLDSPVNAASF GK IIRQFFPQLTTRRLG	120
	* * * * *	
human	T--RGQSKYHYGYIAVKESSQYYDVMYSKKGAAWVSETGKKEVSKQTVAYSPRSKLGTLL	178
mouse	TGTRGQSKYHYGYIAVKESSQYYDVMYSKKGAAWVSETGKREVTKQTVAYSPRSKLGTL	180
zebrafish	T--RGQSKYHYGYIAVKESSQYYDVMYSKKGAAWVNETGK-----	158
	* * * * *	

FIG. 5 Alignment of human and mouse RFX4_v3

human	MHCGLEEPMDSIESWIEPCLNESENKPYSSHTSLGNVSDNENEKENNRASKPHSTPATLQWLEENYEIAEGVCI PRSALYMHYLDCEKNDTQPVNAASFGKIIRQOFPQLTTRPLG	120
mouse	MHCGLEEPMDSIESWIEPCLNESENKPYSSHTSLGNVSDNENEKENNRASKPHSTPATLQWLEENYEIAEGVCI PRSALYMHYLDCEKNDTQPVNAASFGKIIRQOFPQLTTRPLG	120
human	T--RGOSKYYHYGIAVRESSQYVDVMSKGAAMVSETGKPEVSKOTVAYS PRSKLGTLLPEFPNVKDLNLPASLPEEKVSTFIIMYRTHCQILLDTVIRANEDEVQSEFLHFWQGMPPH	238
mouse	TGTFGOSKYYHYGIAVRESSQYVDVMSKGAAMVSETGKPEVSKOTVAYS PRSKLGTLLPEFPNVKDLNLPASLPEEKVSTFIIMYRTHCQILLDTVIRANEDEVQSEFLHFWQGMPPH	240
human	MLPVLGSSTVNVIGVCDISILYAISGVLMPTVLQALPDSLTQVIRKFAKQDEWLAVALHDLHPENLENKFEELSRPESQILKQTSLNHLCOASFTVIHSADITFQMLEDMKNVDLNSI	358
mouse	MLPVLGSSTVNVIGVCDISILYAISGVLMPTVLQALPDSLTQVIRKFAKQDEWLAVALHDLHPENLENKFEELSRPESQILKQTSLNHLCOASFTVIHSADITFQMLEDMKNVDLSSI	360
human	TKQTLTYMEDSDNDEHKPLIQLYQEFDFHLLLEEQSPIESYIEWLDTMVDRCVVVAARKQGSLKVAQQFLMWSQFGRVIFDMTLHSAPSGSEHLLHLMEDDYVLYLLES LHCOEPAN	478
mouse	TKQTLTYMEDSDNDEHKPLIQLYQEFDFHLLLEEQSPIESYIEWLDTMVDRCVVVAARKQGSLKVAQQFLMWSQFGRVIFDMTLHSAPSGSEHLLHLMEDDYVLYLLES LHCOEPAN	480
human	ELMRAMKGEGSTAEVNEEII LTEAAAPTSPVPSPSPAKSATSVEVPPSPSPVSNPSPSEYTGISTGAMQSYTWSLTYYTTAAGSPAENSQQLPCMRNTHVPSSSVTHPIPVYPHKEEH	598
mouse	ELMRAMKGEGSTAEVNEEII LTEATEPTSPGSPSPSPAKSATSVEVPPSPSPVSNPSPSEYTGISTGAMQSYTWSLTYYTTAAGSPAENSQQLPCMRNTHVPSSSVTHPIPVYSHHEEH	600
human	GYTGSYNYGSYGNQHHPHMQSQYPALPHDTAISGELHYAPYHSSAOYPFNSPTSMEPCLMSSTPPLHPTFVTPRWPEVPTANACTSPSVHSTFYGNSSDMYTLTTRNSEYEHMQH	718
mouse	GYTGSYNYGSYGNQHHPHMQSQYPALPHDTAISGELHYAPYHSSAOYPFNSPTSMEPCLMSSTPPLHPTFVTPRWPEVPTANACTSPSVHSTFYGNSSDMYTLTTRNSEYEHMQH	720
human	FPGFAYINGEASTGWAK 735	
mouse	FPGFAYINGEASTGWAK 737	

5/15

6/15

Exon 1

Human MHCGLLEPDMSTESWIERCLNESENKRYSSHTSLGNVNDENEKENNRASKPHSTPA 60
 Mouse MHCGLLEPDMSTESWIERCLNESENKRYSSHTSLGNVNDENEKENNRASKPHSTPA 60
 Zebrafish MLCGLLEPDMSTESWIERCLNESESKRFSSHSSIGNISNDENEKENNRASKPHSTPA 60

Exons 2-5

TLQWLEENYEIAEGVCIPRSALYMHYLDCEKNDTPVNAASFGKIIROQFPOLTRRLG 120
 TLQWLEENYEIAEGVCIPRSALYMHYLDCEKNDTPVNAASFGKIIROQFPOLTRRLG 120
 TLQWLEENYEIAEGVCIPRIALYMHYLDCEKLDSPVNAASFGKIIROQFPOLTRRLG 120

DBD

T--RGQSKYHYGIADVKESSQYIDVMYSKKGAAWVSETGKKEVSKQTVAYSPPRSKLGTL 178
 TGRGQSKYHYGIADVKESSQYIDVMYSKKGAAWVSETGKREVTQTVAYSPPRSKLGTL 180
 T--RGQSKYHYGIADVKESSQYIDVMYSKKGAAWVNETGKKEVTQTVAYSPPRSKLGTL 178

REFPNVKDINGPASTPEEKVSTEMMYRTHCQRLDVTIRANFDEVQSFLHFWGMPPH 238
 PDEPNVKDINGPASTPEEKVSTEMMYRTHCQRLDVTIRANFDEVQSFLHFWGMPPH 240
 PDEPNVKDINGPASTPEEKVSTEMMYRTHCQRLDVTIRANFDEVQSFLHFWGMPPH 238

B

MLPVLCSSSTVNVNIVGVCDSTLYKATSGVLMPTVLOALEDSLTQVIRKFAKQDEWLKVAL 298
 MLPVLCSSSTVNVNIVGVCDSTLYKATSGVLMPTVLOALEDSLTQVIRKFAKQDEWLKVAL 300
 MLPVLCSSSTVNVNIVGVCDSTLYKATSGVLMPTVLOALEDSLTQVIRKFAKQDEWLKVAL 298

C

Exons 6-15

HDLPEENLNRIKFEISRFQSILRRQTSLNHLCOASRTVHSADITFQMLEDWNRVNLNSI 358
 HDLPEENLNRIKFEISRFQSILRRQTSLNHLCOASRTVHSADITFQMLEDWNRVNLSSI 360
 HDLPEENLNRIKFEISRFQSILRRQTSLNHLCOASRTVHSADITFQMLEDWNRVNLNSI 358

TKQTLTYMEDSRDEHRRLTQLYQDFPHLLEEQSPIESYIEWLDMVDRCVVKAAKROG 418
 TKQTLTYMEDSRDEHRRLTQLYQDFPHLLEEQSPIESYIEWLDMVDRCVVKAAKROG 420
 TKQTLTYMEDSRDEHRRLTQLYQDFPHLLEEQSPIESYIEWLDMVDRCVVKAAKROG 418

DD

SLKQVACQELLMSCECTRVIRDMTHSAPSGSGFHLIHMFDVYLYLLESILCOERAN 478
 SLKQVACQELLMSCECTRVIRDMTHSAPSGSGFHLIHMFDVYLYLLESILCOERAN 480
 SLKQVACQELLMSCECTRVIRDMTHSAPSGSGFHLIHMFDVYLYLLESILCOERAN 478

EIMRAMKGECSAEVREETLLEAAATPSVPVSFSPAKSATSMEVPPSSPVSNPSEY 538
 EIMRAMKGECSAEVREETLLEAAATPSVPVSFSPAKSATSMEVPPSSPVSNPSEY 540
 EIMRAMKGECSAEVREETLLEAAATPSVPVSFSPAKSATSMEVPPSSPVSNPSEY 537

TGLS--TGAMQSYTWSLTYYTVAAGSPAENSQQLPCMRN--THVPSSSVTHRIPVYPHRE 596
 TGLS--TGAMQSYTWSLTYYTVAAGSPAENSQQLPCMRN--THVPSSSVTHRIPVYPHRE 598
 TSISATTGAVQSYTWSLTYYTVAAGSPAENSQQLPCMRN--THVPSSSVTHRIPVYPHRE 597

Exons 16-18

EHGYTGSYNYGSYGNQHPHMQSQYPALPHDTAISGPLHYAPYHRSSAQYFPNSPTSME 656
 EHGYTGSYNYGSYGNQHPHMQSQYPALPHDTAISGPLHYAPYHRSSAQYFPNSPTSME 658
 EHGYTGSYNYGSYGNQHPHMQSQYPALPHDTAISGPLHYAPYHRSSAQYFPNSPTSME 657

PCLMSSTPRLHPTPVTPRWPEVPSANTCYTSPSVHSARYGNSSDMYPLTTRNSEYEHEM 716
 PCLMSSTPRLHPTPVTPRWPEVPSANTCYTSPSVHSARYGNSSDMYPLTTRNSEYEHEM 718
 SCLMSGSPILLHSSPVTPRWPEVPSANTCYTSPSVHSARYGNSSDMYPLTTRNSEYEHEM 716

QHFPGFAYINGEASTGWAK 735
 QHFPGFAYINGEASTGWAK 737
 QHFPGFAYINGEASTGWAK 735

FIG. 6

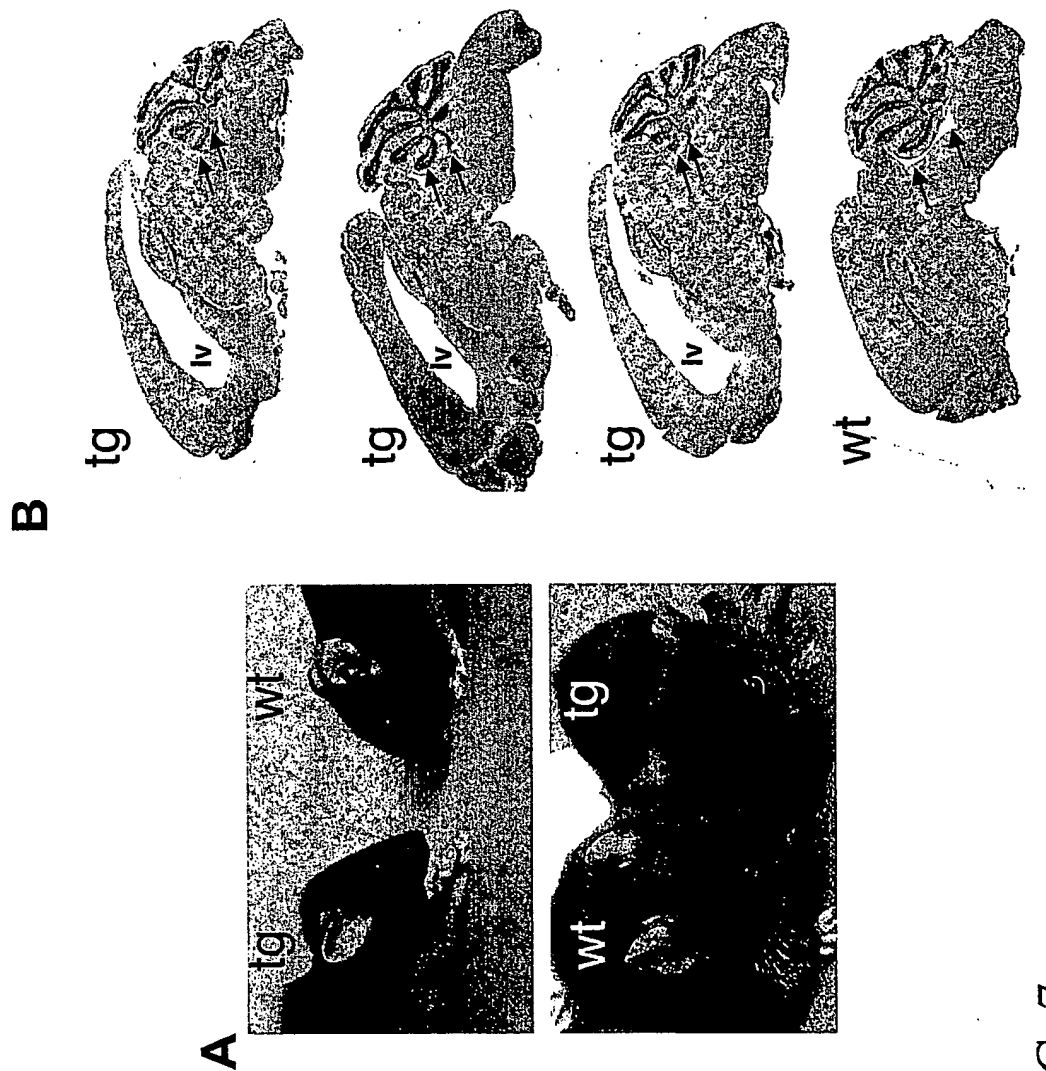


FIG. 7

9/15

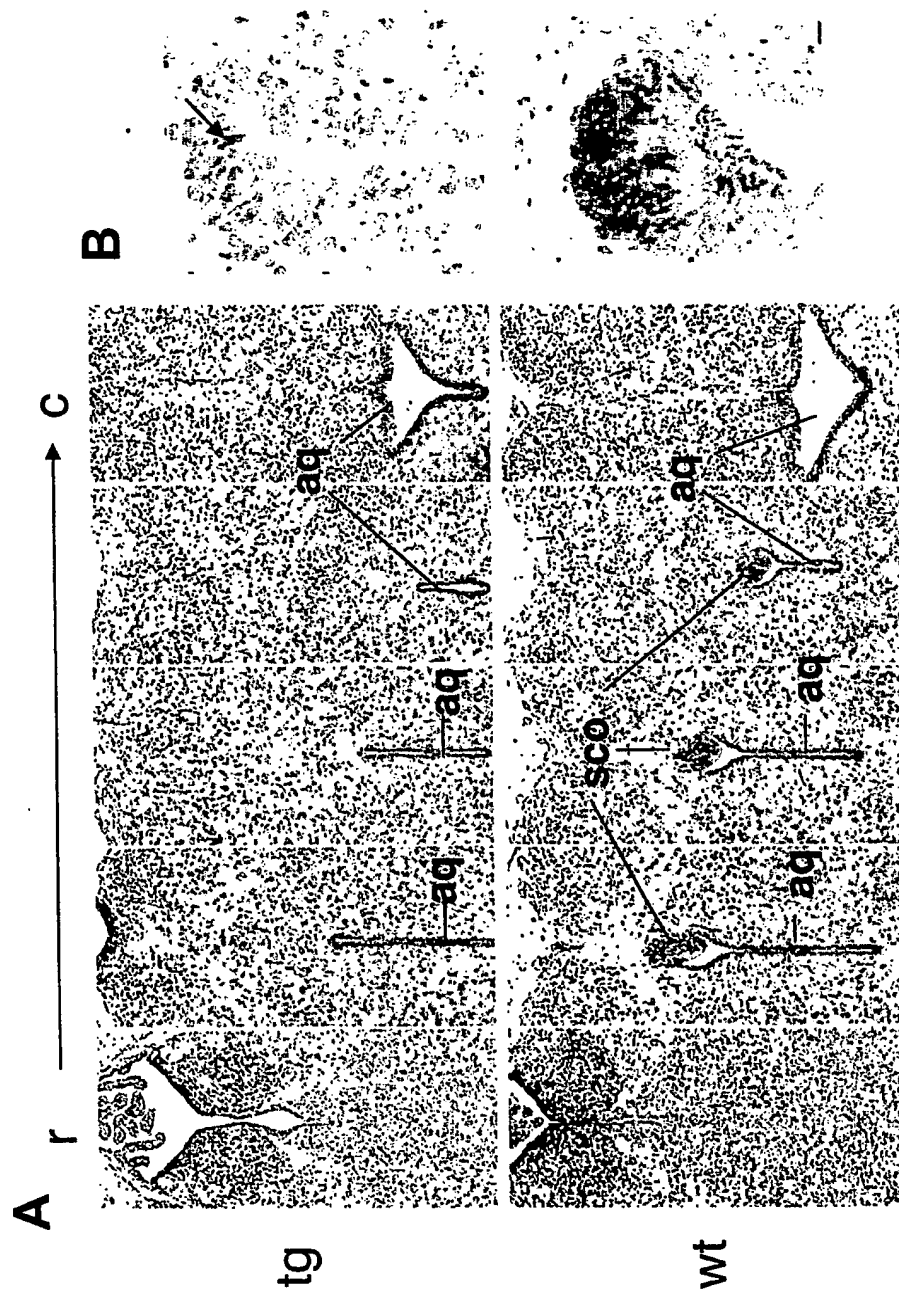


FIG. 9

10/15

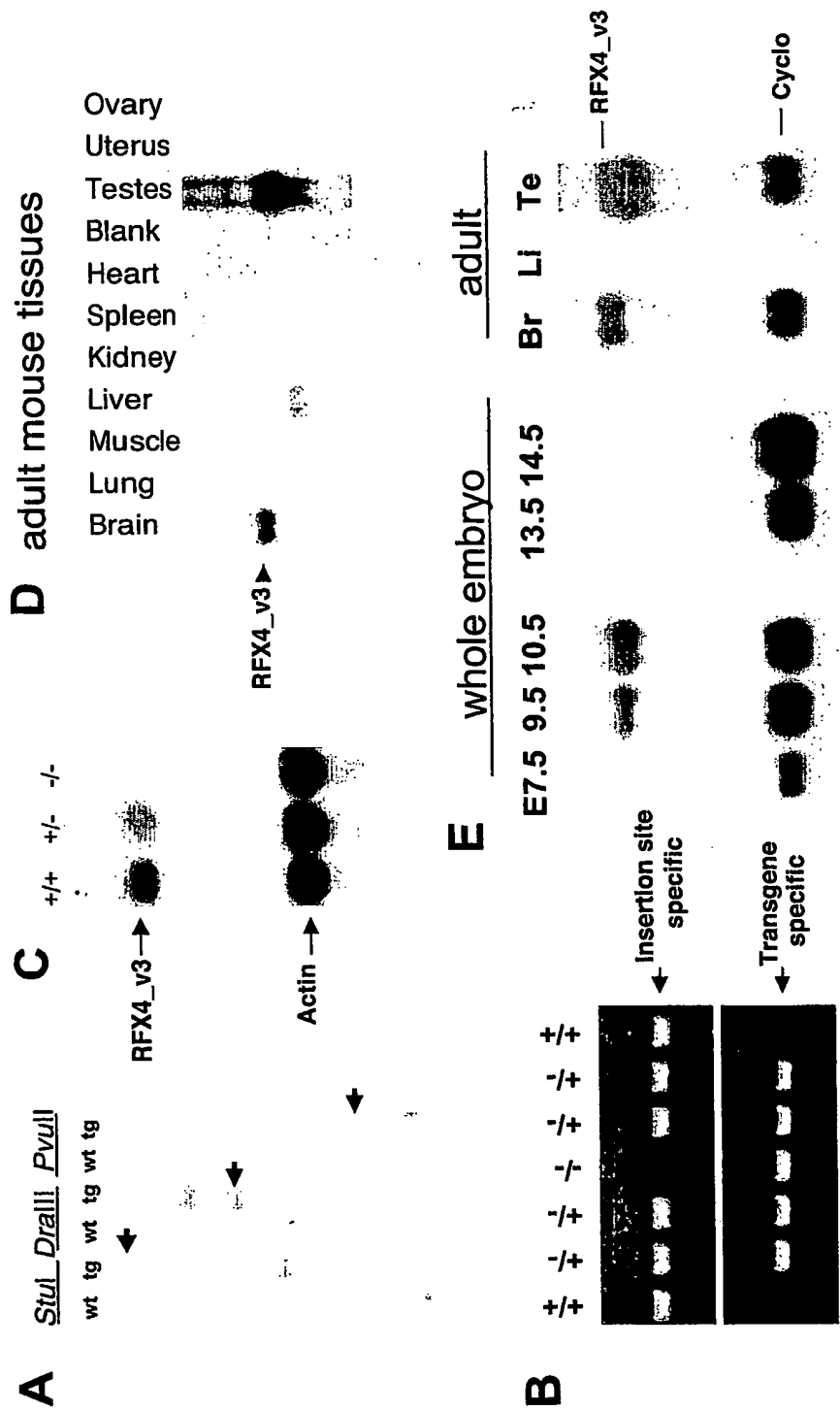


FIG. 10

11/15



FIG. 11

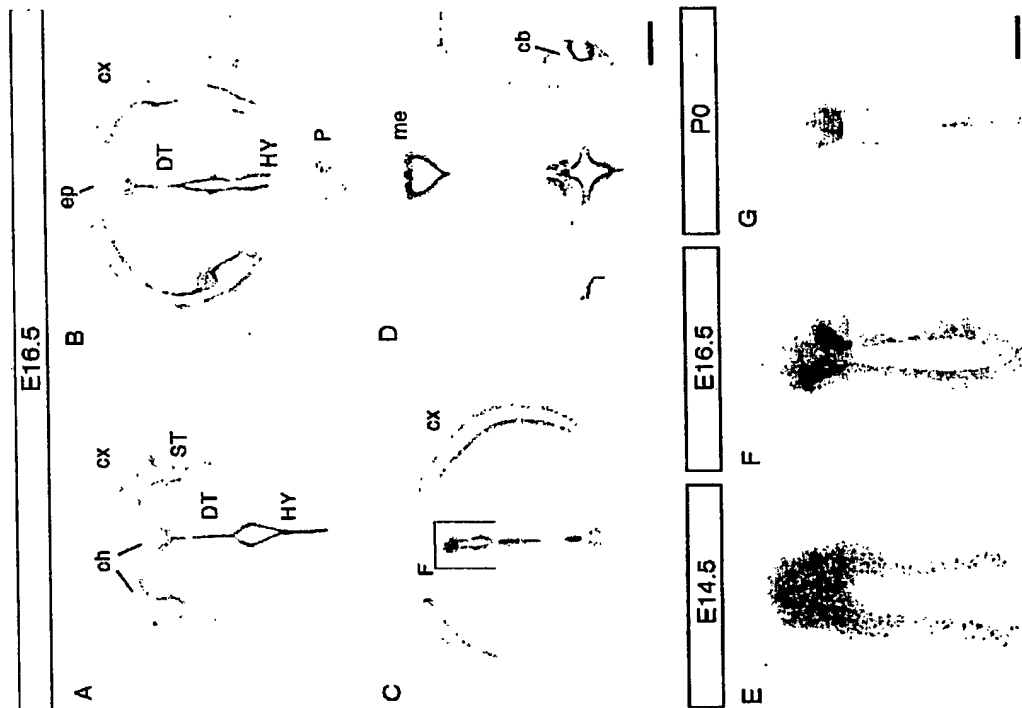


FIG. 13

14/15

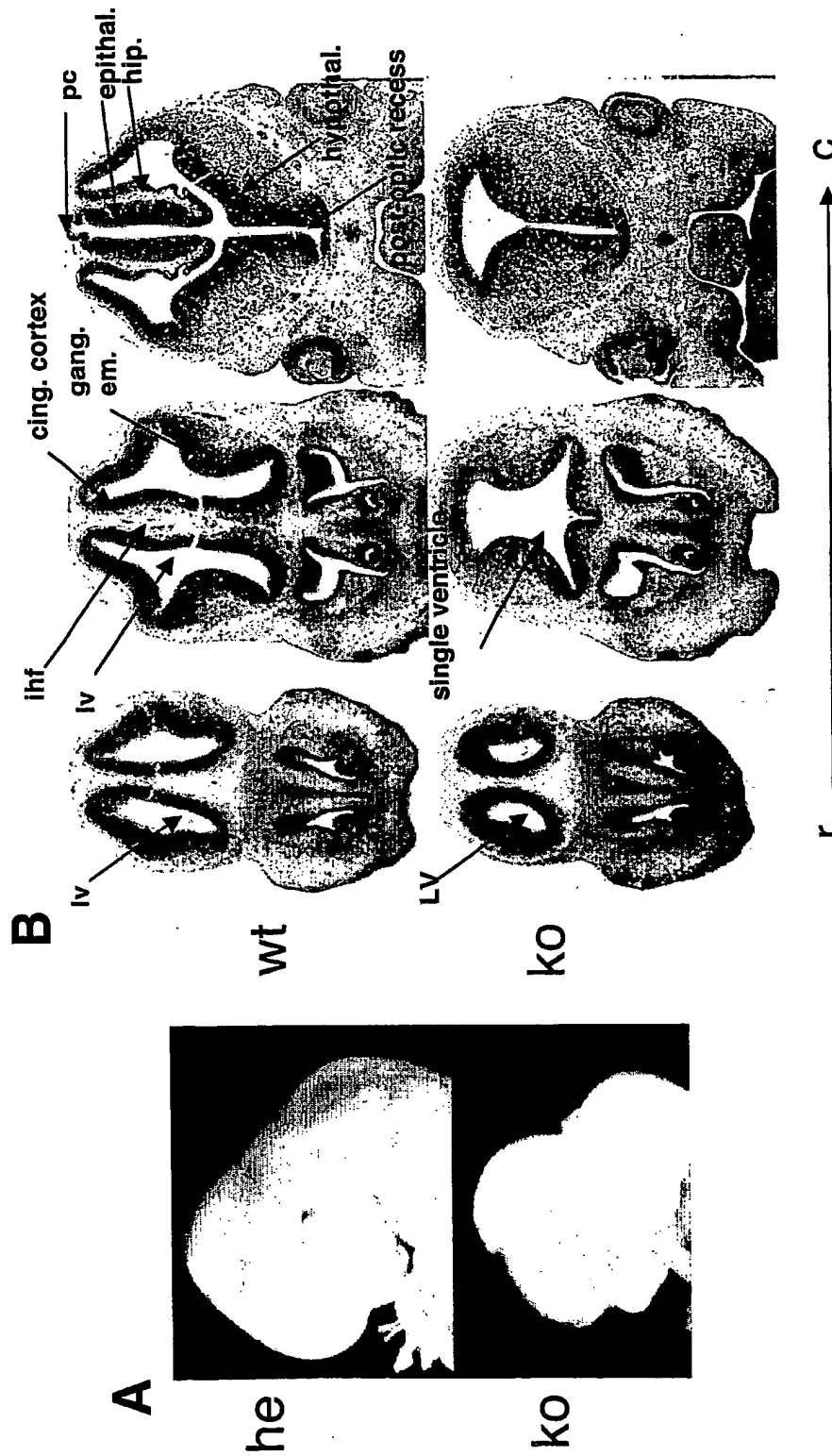


FIG. 14



**This Page is Inserted by IFW Indexing and Scanning
Operations and is not part of the Official Record**

BEST AVAILABLE IMAGES

Defective images within this document are accurate representations of the original documents submitted by the applicant.

Defects in the images include but are not limited to the items checked:

- ☐ BLACK BORDERS
- ☐ IMAGE CUT OFF AT TOP, BOTTOM OR SIDES
- ☒ FADED TEXT OR DRAWING
- ☐ BLURRED OR ILLEGIBLE TEXT OR DRAWING
- ☐ SKEWED/SLANTED IMAGES
- ☐ COLOR OR BLACK AND WHITE PHOTOGRAPHS
- ☐ GRAY SCALE DOCUMENTS
- ☐ LINES OR MARKS ON ORIGINAL DOCUMENT
- ☐ REFERENCE(S) OR EXHIBIT(S) SUBMITTED ARE POOR QUALITY
- ☐ OTHER: _____

IMAGES ARE BEST AVAILABLE COPY.

As rescanning these documents will not correct the image problems checked, please do not report these problems to the IFW Image Problem Mailbox.